Hemoglobinopathies (Sickle Cell)

There are over 600 hemoglobin variants, however only a few are clinically significant. The clinically significant hemoglobinopathies are inherited autosomal recessive disorders of the adult β-hemoglobin chain. Children with two abnormal β-globin genes (homozygotes or double heterozygotes) have a hemoglobin disease whereas those with only one abnormal gene are said to have a hemoglobin trait, which is only of genetic significance. The purpose of newborn screening for hemoglobinopathies is to prevent deaths from pneumococcal sepsis by instituting penicillin prophylaxis and to prevent deaths from splenic sequestration through parent education.

Estimated Incidence (MI):	1 in 600 African American newborns Also seen in individuals of Mediterranean, Indian and Middle Eastern heritage.
Laboratory Screening Test:	Primary test is High Performance Liquid Chromatography (HPLC), which detects abnormal hemoglobins including S, C, D and E. Secondary method is isoelectric focusing (IEF)
Timing of Test:	Valid at birth
Feeding Effect:	None
Transfusion Effect:	The test is invalid on transfused infants. Transfusion may cause false negative or false positive results. **Obtain newborn screen before transfusion.** A retest is required 3 months after the most recent transfusion for all infants not screened before transfusion.
Confirmation:	Infants with positive hemoglobin screening tests are referred to the Sickle Cell Disease Association of America (SCDAA), Michigan Chapter (313) 864-4406 for confirmatory diagnosis and follow up.
Treatment:	Penicillin prophylaxis should begin as soon as possible and continue until six years of age.
Comment:	**The purpose of the newborn hemoglobinopathy-screening program is to identify infants with sickle cell related conditions. Therefore, initial test results that are sickle cell related are designated as positive, and all other results are considered negative. The responsibility for the follow-up of infants found to have non sickle cell related hemoglobinopathies will be left to the discretion of the physician of record.**

For further information, contact the Newborn Screening Program
Telephone (517) 335-9205
Email mdch-newbornscreening@michigan.gov

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